

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

**Table 2. FEVR Mutations Detected in *FZD4***

Location	cDNA change	Protein change	Publication	Phenotype
Exon 1	97C→T	P33S	MacDonald et al 2005, Nallathambi et al 2006	adFEVR
Exon 1	107G>A	G36D	Toomes, Bottomley, Scott et al 2004	adFEVR
<i>Exon 1**</i>	<i>205C&gt;T</i>	<i>H69Y</i>	<i>Kondo et al 2003, Omoto et al 2004</i>	adFEVR
Exon 1	244-251del8ins27	F82fsX135	Nallathambi et al 2006	adFEVR
Exon 2	313A>G	M105V	Kondo et al 2003, Xu et al 2004	adFEVR
Exon 2	314T>C	M105T	Toomes, Bottomley, Scott et al 2004	adFEVR
Exon 2	469A>G	M157V	Toomes, Bottomley, Scott et al 2004; Xu et al 2004	adFEVR
<i>Exon 2**</i>	<i>502C&gt;T</i>	<i>P168S</i>	Toomes, Bottomley, Scott et al 2004; <i>MacDonald et al 2005</i>	adFEVR
Exon 2	541T>C	C181R	Omoto et al 2004	adFEVR
Exon 2	610T>C	C204R	Nallathambi et al 2006	adFEVR
Exon 2	678G>A	W226X	Jiang et al 2004	adFEVR
Exon 2	957delG	W319fsX323	Toomes, Bottomley, Scott et al 2004	adFEVR
Exon 2	957G>A	W319X	Kondo et al 2003, Gal et al 2004	adFEVR
Exon 2	1005G>C	W335C	Qin et al 2005	adFEVR
Exon 2	1024A>G	M342V	Qin et al 2005, Yoshida et al 2004	adFEVR
Exon 2	1250G>C*	R417Q*	Kondo et al 2003	adFEVR
Exon 2	1282del4	D428fsX429	Gal et al 2004	adFEVR
Exon 2	1286del5	K429fsX456	Gal et al 2004	adFEVR
Exon 2	1363A>C	T445P	Gal et al 2004	adFEVR
Exon 2	1463G>A	G488D	Kondo et al 2003	adFEVR
Exon 2	1474G>C	G492R	Gal et al 2004	adFEVR
Exon 2	unknown	W496X	Gal et al 2004	adFEVR
Exon 2	1490C>T	S497F	Toomes, Bottomley, Scott et al 2004	adFEVR
Exon 2	1479-1484delGTGGAT	M493-W494del	Robitaille et al 2002	adFEVR
Exon 2	1498delA	T500fsX512	Toomes, Bottomley, Scott et al 2004	adFEVR
Exon 2	1501-1502delCT	L501fsX533	Robitaille et al 2002; Toomes, Bottomley, Scott et al 2004	adFEVR
Exon 2	1513C>T	Q505X	Toomes, Bottomley, Scott et al 2004	adFEVR

\* A second mutation in *LRP5* (R444C) also reported in this family [Qin et al 2005]

\*\* The pathogenic nature of the two mutations in italics is questionable due to the detection of these changes in control individuals (healthy volunteers who have not undergone fundi examination). H69Y was present in 2/300 control chromosomes but was also found as a second *FZD4* change in a severely affected member of a family with a G488D mutation [Kondo et al 2003]. It was also found in a sporadic FEVR patient (no further family members available for testing) [Kondo et al 2003] and in a small family with three affected members [Omoto et al 2004]. Similarly, P168S was detected in 1/400 control chromosomes but was also found in a sporadic FEVR patient [Toomes et al 2004, unpublished observation]. It was also found on the same allele as P33S in a sporadic patient [MacDonald et al 2005].